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<110> ML4 Foundation
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Acierno, James S.

<120> A Gene Encoding A New TRP Channel is Mutated in Mucopolipidosis IV

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<140> 09/851,494

<141> 2001-05-08

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cccgaaatttc						13270

<210> 2
 <211> 2051
 <212> DNA
 <213> Homo sapiens

<400> 2	
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ccagcatgac	agccccggcg ggtcgcgcgc gctcagagac cgagcggtt ctgaccccca 180
acccccgggt	atgggacccag gcggggcctt caccggcccc tccgacaccc ccagaagagg 240
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tcgccttccg	acacctcttc ctgctgggct actcggacgg agcggatgac accttcgcag 480
cctacacgcg	ggagcagctg taccaggcca tcttccatgc tgtggaccag tacctggcgt 540
tgcttgacgt	gtcactgggc cggatatgct atgtccgtgg tgggggtgac ccttggacca 600
atggctcagg	gcttgccttc tgccagcggc actaccaccg aggccacgtg gacccggcca 660
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agaacctcac gctcaaattc cacaagctgg tcaatgtcac catccacttc cggctgaaga	840
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cggcctgcag ccttctctgc tgctgcggaa gggacccctc ggaggagcat tcgctgctgg	1860
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cccgaccccg cttattttatt tgtagggttt gcttttaagg atcggctccc tgtcgcgccc	1980
gaggagggcc tggacctttc gtgtcggacc cttggggggcg gggagactgg gtggggaggg	2040
tgttgaataa a	2051

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 <211> 580
 <212> PRT
 <213> Homo sapiens

<400> 3

Met Thr Ala Pro Ala Gly Pro Arg Gly Ser Glu Thr Glu Arg Leu Leu
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Thr Pro Asn Pro Gly Tyr Gly Thr Gln Ala Gly Pro Ser Pro Ala Pro
20 25 30

Pro Thr Pro Pro Glu Glu Glu Asp Leu Arg Arg Arg Leu Lys Tyr Phe
35 40 45

Phe Met Ser Pro Cys Asp Lys Phe Arg Ala Lys Gly Arg Lys Pro Cys
50 55 60

Lys Leu Met Leu Gln Val Val Lys Ile Leu Val Val Thr Val Gln Leu
65 70 75 80

Ile Leu Phe Gly Leu Ser Asn Gln Leu Ala Val Thr Phe Arg Glu Glu
85 90 95

Asn Thr Ile Ala Phe Arg His Leu Phe Leu Leu Gly Tyr Ser Asp Gly
100 105 110

Ala Asp Asp Thr Phe Ala Ala Tyr Thr Arg Glu Gln Leu Tyr Gln Ala
115 120 125

Ile Phe His Ala Val Asp Gln Tyr Leu Ala Leu Pro Asp Val Ser Leu
130 135 140

Gly Arg Tyr Ala Tyr Val Arg Gly Gly Gly Asp Pro Trp Thr Asn Gly
145 150 155 160

Ser Gly Leu Ala Leu Cys Gln Arg Tyr Tyr His Arg Gly His Val Asp
165 170 175

Pro Ala Asn Asp Thr Phe Asp Ile Asp Pro Met Val Val Thr Asp Cys
180 185 190

Ile Gln Val Asp Pro Pro Glu Arg Pro Pro Pro Pro Pro Ser Asp Asp
195 200 205

Leu Thr Leu Leu Glu Ser Ser Ser Ser Tyr Lys Asn Leu Thr Leu Lys
210 215 220

Phe His Lys Leu Val Asn Val Thr Ile His Phe Arg Leu Lys Thr Ile
225 230 235 240

Asn Leu Gln Ser Leu Ile Asn Asn Glu Ile Pro Asp Cys Tyr Thr Phe
245 250 255

Ser Val Leu Ile Thr Phe Asp Asn Lys Ala His Ser Gly Arg Ile Pro
260 265 270

Ile Ser Leu Glu Thr Gln Ala His Ile Gln Glu Cys Lys His Pro Ser
275 280 285

Val Phe Gln His Gly Asp Asn Ser Phe Arg Leu Leu Phe Asp Val Val
290 295 300

Val Ile Leu Thr Cys Ser Leu Ser Phe Leu Leu Cys Ala Arg Ser Leu
305 310 315 320

Leu Arg Gly Phe Leu Leu Gln Asn Glu Phe Val Gly Phe Met Trp Arg
325 330 335

Gln Arg Gly Arg Val Ile Ser Leu Trp Glu Arg Leu Glu Phe Val Asn
340 345 350

Gly Trp Tyr Ile Leu Leu Val Thr Ser Asp Val Leu Thr Ile Ser Gly
355 360 365

Thr Ile Met Lys Ile Gly Ile Glu Ala Lys Asn Leu Ala Ser Tyr Asp
370 375 380

Val Cys Ser Ile Leu Leu Gly Thr Ser Thr Leu Leu Val Trp Val Gly
385 390 395 400

Val Ile Arg Tyr Leu Thr Phe Phe His Asn Tyr Asn Ile Leu Ile Ala
405 410 415

Thr Leu Arg Val Ala Leu Pro Ser Val Met Arg Phe Cys Cys Cys Val
420 425 430

Ala Val Ile Tyr Leu Gly Tyr Cys Phe Cys Gly Trp Ile Val Leu Gly
435 440 445

Pro Tyr His Val Lys Phe Arg Ser Leu Ser Met Val Ser Glu Cys Leu
450 455 460

Phe Ser Leu Ile Asn Gly Asp Asp Met Phe Val Thr Phe Ala Ala Met
465 470 475 480

Gln Ala Gln Gln Gly Arg Ser Ser Leu Val Trp Leu Phe Ser Gln Leu
485 490 495

Tyr Leu Tyr Ser Phe Ile Ser Leu Phe Ile Tyr Met Val Leu Ser Leu
500 505 510

Phe Ile Ala Leu Ile Thr Gly Ala Tyr Asp Thr Ile Lys His Pro Gly
515 520 525

Gly Ala Gly Ala Glu Glu Ser Glu Leu Gln Ala Tyr Ile Ala Gln Cys
530 535 540

Gln Asp Ser Pro Thr Ser Gly Lys Phe Arg Arg Gly Ser Gly Ser Ala
545 550 555 560

Cys Ser Leu Leu Cys Cys Cys Gly Arg Asp Pro Ser Glu Glu His Ser
565 570 575

Leu Leu Val Asn
580

<211> 553
<212> PRT
<213> Homo sapiens

<400> 4

Met Ala Asp Pro Glu Val Val Val Cys Ser Cys Ser Ser His Glu Glu
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Glu Asn Arg Cys Asn Phe Asn Gln Gln Thr Ser Pro Ser Glu Glu Leu
20 25 30

Leu Leu Glu Asp Gln Met Arg Arg Lys Leu Lys Phe Phe Phe Met Asn
35 40 45

Pro Cys Glu Lys Phe Trp Ala Arg Gly Arg Lys Pro Trp Lys Leu Ala
50 55 60

Ile Gln Ile Leu Lys Ile Ala Met Val Thr Ile Gln Leu Val Leu Phe
65 70 75 80

Gly Leu Ser Asn Gln Met Val Val Ala Phe Lys Glu Glu Asn Thr Ile
85 90 95

Ala Phe Lys His Leu Phe Leu Lys Gly Tyr Met Asp Arg Met Asp Asp
100 105 110

Thr Tyr Ala Val Tyr Thr Gln Ser Asp Val Tyr Asp Gln Leu Ile Phe
115 120 125

Ala Val Asn Gln Tyr Leu Gln Leu Tyr Asn Val Ser Val Gly Asn His
130 135 140

Ala Tyr Glu Asn Lys Gly Thr Lys Gln Ser Ala Met Ala Ile Cys Gln
145 150 155 160

His Phe Tyr Lys Arg Gly Asn Ile Tyr Pro Gly Asn Asp Thr Phe Asp
165 170 175

Ile Asp Pro Glu Ile Glu Thr Glu Cys Phe Phe Val Glu Pro Asp Glu
180 185 190

Pro Phe His Ile Gly Thr Pro Ala Glu Asn Lys Leu Asn Leu Thr Leu
195 200 205

Asp Phe His Arg Leu Leu Thr Val Glu Leu Gln Phe Lys Leu Lys Ala
210 215 220

Ile Asn Leu Gln Thr Val Arg His Gln Glu Leu Pro Asp Cys Tyr Asp
225 230 235 240

Phe Thr Leu Thr Ile Thr Phe Asp Asn Lys Ala His Ser Gly Arg Ile
245 250 255

Lys Ile Ser Leu Asp Asn Asp Ile Ser Ile Arg Glu Cys Lys Asp Trp
260 265 270

His Val Ser Gly Ser Ile Gln Lys Asn Thr His Tyr Met Met Ile Phe
275 280 285

Asp Ala Phe Val Ile Leu Thr Cys Leu Val Ser Leu Ile Leu Cys Ile
290 295 300

Arg Ser Val Ile Arg Gly Leu Gln Leu Gln Gln Glu Phe Val Asn Phe
305 310 315 320

Phe Leu Leu His Tyr Lys Lys Glu Val Ser Val Ser Asp Gln Met Glu
325 330 335

Phe Val Asn Gly Trp Tyr Ile Met Ile Ile Ile Ser Asp Ile Leu Thr
340 345 350

Ile Ile Gly Ser Ile Leu Lys Met Glu Ile Gln Ala Lys Ser Leu Thr
355 360 365

Ser Tyr Asp Val Cys Ser Ile Leu Leu Gly Thr Ser Thr Met Leu Val
370 375 380

Trp Leu Gly Val Ile Arg Tyr Leu Gly Phe Phe Ala Lys Tyr Asn Leu
385 390 395 400

Leu Ile Leu Thr Leu Gln Ala Ala Leu Pro Asn Val Ile Arg Phe Cys
405 410 415

Cys Ala Ala Met Ile Tyr Leu Gly Tyr Cys Phe Cys Gly Trp Ile
420 425 430

Val Leu Gly Pro Tyr His Asp Lys Phe Arg Ser Leu Asn Met Val Ser
435 440 445

Glu Cys Leu Phe Ser Leu Ile Asn Gly Asp Asp Met Phe Ala Thr Phe
450 455 460

Ala Lys Met Gln Gln Lys Ser Tyr Leu Val Trp Leu Phe Ser Arg Ile
465 470 475 480

Tyr Leu Tyr Ser Phe Ile Ser Leu Phe Ile Tyr Met Ile Leu Ser Leu
485 490 495

Phe Ile Ala Leu Ile Thr Asp Thr Tyr Glu Thr Ile Lys Gln Tyr Gln
500 505 510

Gln Asp Gly Phe Pro Glu Thr Glu Leu Arg Thr Phe Ile Ser Glu Cys
515 520 525

Lys Asp Leu Pro Asn Ser Gly Lys Tyr Arg Leu Glu Asp Asp Pro Pro
530 535 540

Val Ser Leu Phe Cys Cys Cys Lys Lys
545 550

<210> 5
<211> 652
<212> PRT
<213> Drosophila

<400> 5

Met Gln Ser Tyr Gly Pro Gly Ala Gln Thr Ala Pro Ala Val Lys Arg
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Arg Thr Asp Ser Tyr Glu Ala Ala Gln Gln Gln Gln Ser Pro Glu
20 25 30

Ser Asp Glu Glu Tyr Val Asn Thr Arg Ile Leu Arg Arg Gln Val Gln
35 40 45

Leu Gln Ser Thr Pro Val Ala Pro Val Val Pro Met Pro Ile Ser Ala
50 55 60

Gly Ser Gly Thr Ala Pro Pro Ser Val Asp Gly Arg Glu Glu Gln Pro
65 70 75 80

Glu Phe Pro Gly Ser Ser Ala Ala Ser Tyr Gln Glu Glu Arg Met Arg
85 90 95

Arg Lys Leu Gln Phe Phe Phe Met Asn Pro Ile Glu Lys Trp Gln Ala
100 105 110

Lys Arg Lys Phe Pro Tyr Lys Phe Val Val Gln Ile Val Lys Ile Phe
115 120 125

Leu Val Thr Met Gln Leu Cys Leu Phe Ala His Ser Arg Tyr Asn His
130 135 140

Ile Asn Tyr Thr Gly Asp Asn Arg Phe Ala Phe Ser His Leu Phe Leu
145 150 155 160

Arg Gly Trp Asp Ser Ser Arg Glu Val Glu Ser Tyr Pro Pro Ala Val
165 170 175

Gly Pro Phe Ala Leu Tyr Leu Lys Ser Glu Phe Phe Asp Thr Val Gln
180 185 190

Tyr Ala Val Asn Gly Tyr Ala Asn Val Ser Arg Ser Ile Gly Pro Tyr
195 200 205

Asp Tyr Pro Thr Pro Asn Asn Thr Met Pro Pro Leu Lys Leu Cys Leu
210 215 220

Gln Asn Tyr Arg Glu Gly Thr Ile Phe Gly Phe Asn Glu Ser Tyr Ile
225 230 235 240

Phe Asp Pro His Ile Asp Glu Val Cys Glu Arg Leu Pro Pro Asn Val
245 250 255

Thr Thr Ile Gly Val Glu Asn Tyr Leu Arg Gln Arg Asp Val Glu Val
260 265 270

Asn Phe Ala Ser Leu Val Ser Ala Gln Leu Thr Phe Lys Ile Lys Thr
275 280 285

Val Asn Phe Lys Ala Asn Gly Gly Pro Leu Ser Ala Pro Asp Cys Phe
290 295 300

Arg Phe Asp Ile Ser Ile Thr Phe Asn Asn Arg Asp His Asp Gly Gln
305 310 315 320

Met Leu Leu Ser Leu Asp Ala Glu Ala Thr Arg Leu Lys Cys His Gly
325 330 335

Ala Thr Asp Phe Ile Ser Asp Ala Asn Phe Asp Ser Met Leu Arg Ser
340 345 350

Val Leu Asn Ile Phe Val Leu Leu Thr Cys Ala Leu Ser Phe Ala Leu
355 360 365

Cys Thr Arg Ala Leu Trp Arg Ala Tyr Leu Leu Arg Cys Thr Thr Val
370 375 380

Asn Phe Phe Arg Ser Gln Phe Gly Lys Glu Leu Ser Phe Asp Gly Arg
385 390 395 400

Leu Glu Phe Val Asn Phe Trp Tyr Ile Met Ile Ile Phe Asn Asp Val

405

410

415

Leu Leu Ile Ile Gly Ser Ala Leu Lys Glu Gln Ile Glu Gly Arg Tyr
 420 425 430

Leu Val Val Asp Gln Trp Asp Thr Cys Ser Leu Phe Leu Gly Ile Gly
 435 440 445

Asn Leu Leu Val Trp Phe Gly Val Leu Arg Tyr Leu Gly Phe Phe Lys
 450 455 460

Thr Tyr Asn Val Val Ile Leu Thr Leu Lys Lys Ala Ala Pro Lys Ile
 465 470 475 480

Leu Arg Phe Leu Ile Ala Ala Leu Leu Ile Tyr Ala Gly Phe Val Phe
 485 490 495

Cys Gly Trp Leu Ile Leu Gly Pro Tyr His Met Lys Phe Arg Ser Leu
 500 505 510

Ala Thr Thr Ser Glu Cys Leu Phe Ala Leu Ile Asn Gly Asp Asp Met
 515 520 525

Phe Ala Thr Phe Ala Thr Leu Ser Ser Lys Ala Thr Trp Leu Trp Trp
 530 535 540

Phe Cys Gln Ile Tyr Leu Tyr Ser Phe Ile Ser Leu Tyr Ile Tyr Val
 545 550 555 560

Val Leu Ser Leu Phe Ile Ala Val Ile Met Asp Ala Tyr Asp Thr Ile
 565 570 575

Lys Ala Tyr Tyr Lys Asp Gly Phe Pro Thr Thr Asp Leu Lys Ala Phe
 580 585 590

Val Gly Thr Arg Thr Ala Glu Asp Ile Ser Ser Gly Val Phe Met Thr
 595 600 605

Asp Leu Asp Asp Phe Asp Gln Thr Ser Phe Leu Asp Val Val Lys Ser
610 615 620

Ile Cys Cys Cys Gly Arg Cys Gly Arg His Gln Glu Pro Ala Gln Pro
625 630 635 640

Asn Ser Gly Tyr Thr Ser Leu Ser Ser Ile Met Lys
645 650

<210> 6
<211> 26
<212> DNA
<213> Artificial Sequence

<220>
<223> PCR primer

<400> 6
cgagggagcg aggtcgagc gacagc

26

<210> 7
<211> 25
<212> DNA
<213> Artificial Sequence

<220>
<223> PCR primer

<400> 7
aacaccctcc ccacccagtc tcccc

25

<210> 8
<211> 21
<212> DNA
<213> Artificial Sequence

<220>
<223> PCR primer

<400> 8
caacctctac taccctctcc c

21

<210> 9
<211> 18

<212> DNA
<213> Artificial Sequence

<220>
<223> PCR primer

<400> 9
aacagtgaag cctcgtcc

18

<210> 10
<211> 23
<212> DNA
<213> Artificial Sequence

<220>
<223> PCR primer

<400> 10
gatataaatg gcaggcagct ttc

23

<210> 11
<211> 20
<212> DNA
<213> Artificial Sequence

<220>
<223> PCR primer

<400> 11
ctcaccgtgc tggaagacac

20